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Archives

Intraspecific chromosome variability

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Abstract. (Editorial preface). The publication is presented in order to remind us of one of dramatic pages of the history of genetics. It re-opens for the contemporary reader a comprehensive work marking the priority change from plant cytogenetics to animal cytogenetics led by wide population studies which were conducted on *Drosophila* polytene chromosomes. The year of the publication (1937) became the point of irretrievable branching between the directions of Old World and New World genetics connected with the problems of chromosome variability and its significance for the evolution of the species. The famous book of T. Dobzhansky (1937) was published by Columbia University in the US under the title "Genetics and the origin of species", and in the shadow of this American 'skybuilding' all other works grew dim. It is remarkable that both Dobzhansky and Dubinin come to similar conclusions about the role of chromosomes in speciation. This is not surprising given that they both might be considered as representatives of the Russian genetic school, by their birth and education. Interestingly, Dobzhansky had never referred to the full paper of Dubinin et al. (1937), though a previous short communication in *Nature* (1936) was included together with all former papers on the related subject. In full, the volume of the original publication printed in the *Biological Journal* in Moscow comprised 47 pages, in that number 41 pages of the Russian text accompanied by 16 Figs, a table and reference list, and, above all, 6 pages of the English summary. This final part in English is now reproduced in the authors' version with the only addition being the reference list in the originally printed form.

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Contents

Summary

1. Studies of the karyotypes of related species (the most significant investigations have been carried out on nuclei of the salivary gland chromosomes in hybrid *Drosophila* larvae as, for instance, the work of Dobzhansky and Tan, 1936, and other authors) have shown that the fission of species is often accompanied by a complex evolution of the karyotypes. This evolution evidently rests on the successive utilization of separate elementary changes of the chromosome structure, interchromosomal exchanges taking a greater share than intrachromosome rearrangements. So far there has been a sharp gap in our knowledge between the results of the evolution of karyotypes and the factors, which bring about this evolution.

The low frequency for spontaneous change of chromosome structure explains the widespread viewpoint that natural populations possess only isolated rare chromosome changes. A series of Diptera species have been subjected to an extensive investigation by the authors of the present work in the years 1935 – 1936 (see preliminary communication in *Nature*, 1936). These studies have shown that this point of view is erroneous and that chromosomal mutations are a common feature of natural populations.

2. The principal source of material constituted larvae of wild populations. The studies of chromosome complexes were carried out on the giant chromosomes of the salivary gland nuclei of larvae by means of the aceto-carmine method. The following species were used: *D. melanogaster*, *D. funebris*, *D. obscura*, *D. obscura* 2, *D. obscura* 3, two unidentified species (one from Bakuriani, one from Sukhumi) and two *Chironomus* species.

The populations were investigated in the following localities: Central Russia (Moscow, Liublino, the village Polivanovo, Jaroslavl, Zagorsk), North Caucasus (Gelendzhik, Derbent), Transcaucasus (Sukhumi, Batumi, Kutaissi, Gori, Borjom - Bakuriani, Gurdzhani - Tsinandali, Erevan, Baku), Semirechje (Alma-Ata, Talgar), Central Asia (Djalal-Abad, Osh, Fergana, Leninabad, Samarkand, Stalinabad, Termez, Bukhara, Ashkhabad).

The populations of Moscow, Kutaissi, Batumi, Gori, Sukhumi were studied for the first time in 1935 and a second time in 1936.

3. Our studies have shown that chromosome aberrations are widespread in nature. 34 515 chromosomes were investigated in *D. melanogaster*. Eight mutations were found: a deletion of the X-chromosome, one inversion in the left arm of chromosome II, two

inversions in the right arm of chromosome II, four inversions in the right arm of chromosome III[1].

The long IIR inversion and the IIL inversion were oftener found together than apart. Our study has shown that this double inversion is homologous to the inversion CI1L Cy CIIR found by Ward in Michigan. Five inversions were found in *D. funebris*: three in chromosome II, one in III and one in IV. In *D. obscura* one inversion was found in the left arm of chromosome II and a small inversion in the right arm of chromosome II. In an unidentified *Drosophila* species collected in Sukhumi a double inversion involving 3 breaks was found, two breaks having occurred in the active region and one in the inert one. In the Bakuriani species one inversion was found. In *D. obscura* 2 six different inversions and one interchromosomal displacement were found. In. *D. obscura* 3 the following eight mutations were met, four inversions, two rearrangements and two complex aberrations. In one *Chironomus* species four different inversions were found.

The aberrations described here are frequently found in the populations studied, 939 aberrations were found in 42315 chromosomes studied. These mutations are intrachromosomal inversions and displacements with the exception of one inversion in *D. obscura*. All these mutations, which have been genetically studied, are viable in homozygous condition.

4. The geographical distribution of chromosome aberrations has been studied in an extensive area for *D. melanogaster*, *D. obscura* and *D. funebris*. A widespread distribution of single mutations has invariably been found.

The CIIL A inversion of *D. obscura* has been found in the populations of Moscow, Alma-Ata, Samarkand and Gori. The CП Ml inversion of *D. funebris* has been found in the populations of Moscow, Alma-Ata, Djaljal-Abad. The inversion CIIIR G of *D. melanogaster* was met on the coasts of the Black and Caspian seas, throughout Central Asia from Ashkhabad till Stalinabad and Osh. The inversion CIIIR K2 was found in Transcaucasus and Central Asia as well as the CIIIR G inversion, and so on. Homologous aberrations are, therefore, widespread. 34 515 chromosomes were studied in *D. melanogaster*, and 525 chromosomes bearing inversions were found in 20 populations, and they represented only seven different inversions.

In *D. funebris* 207 chromosomes bearing inversions were found among 5517 chromosomes (they contained only five different inversions) in seven populations. Based on the distribution of homologous aberrations definitely regular geographical distributions of separate chromosome mutations were established. There also is a clear and often marked differentiation of the frequencies of mutations in separate populations.

5. The factors determining the frequencies of different mutations in wild populations are represented by the mutability of the chromosomes, the automatic genetic processes (acting in isolated or freely crossing groups) and the action of natural selection.

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[1] Foot-note: The first letter stands for the rearrangement (C -crossing-over inhibitor, i.e. inversion), roman number indicate the chromosome affected, the subsequent letter specifies the chromosome arm (R – right, L – left), the last letter represents the initial of the locality (f.i., M – Moscow, K – Kutaissi); to differentiate the different inversions affecting the same chromosome arm Arabian numbers are added to the initial (K1, K2 represent two inversions in Kutaissi).

Table. Frequencies of various inversions in different populations of *D. melanogaster* (in per cent).

Populations / (q)Frequencies of inversions	Alma-Ata	Djalal-Abad	Osh	Fergana	Leninabad	Samarkand	Bukhara	Stalinabad	Termez	Ashkhabad	Baku	Kakhetia	Gori	Kutaissi	Batumi	Sukhumi	Erevan	Gelendzhik	Derbent
CIIIR G1	—	2.46	0.30	0.24	0.10	2 20	10.94	3.85	16.04	3.54	0.38	—	—	—	—	—	—	7.40	2.1
CIIIR K2	—	—	0.15	5.36	1.89	—	5.68	6.93	—	4.71	—	—	0.68	0.52	—	—	—	—	—
CIIIR K1	—	—	—	—	—	—	—	—	—	—	—	—	—	0.84	—	—	—	—	—
CIIIR S	—	—	—	—	—	0.32	—	—	—	—	—	—	—	—	—	—	—	—	—
CIIR B	—	—	—	—	—	—	—	—	—	—	—	—	—	—	0.91	—	—	—	—
CIIL CIIR K	—	—	—	—	—	—	—	—	—	—	0.58	—	0.42	1.58	4.14	—	—	—	—
CIIR K	—	—	—	—	—	—	—	—	—	—	—	—	0.34	0.10	—	—	—	—	—
CIIL K	—	—	—	—	—	—	—	—	—	—	—	—	0.42	—	—	—	—	—	—

However, the mutation process alone cannot be held responsible for the permeation of species with certain chromosome mutations. Firstly, the mutation rate is not sufficiently high. Secondly, recurrent mutability seems out of the question for it would have had to produce the millions of homologous aberrations present in a given population. Recurrent appearance of a given structural aberration is practically excluded because it differs from the recurrent mutation of genes in that it requires two independent events i. e., the coincidence of at least two changes in the chromosome. All this renders it probable that a great number of representatives of a determined chromosome mutation, widely distributed throughout the area of the species, is the result of a single mutation.

Responsible for the different frequencies of a given aberration in different populations are primarily the automatic genetic processes where there exists isolation. The table gives a summary of the frequencies of chromosome mutations throughout all the populations of *D. melanogaster* studied.

We see that the concentration of the CIIIR G fluctuates in the populations of Central Asia between 16, 0.4 and 0 per cent. The CII Ml inversion in *D. funebris* occupies 15.5 per cent in Moscow, in Djaljal-Abad – 1.25 per cent, in Alma-Ata and Stalinabad – 0 per cent. The population of *D. melanogaster* of Batumi shows a significant frequency shift throughout the year. The CIIL - CIIR K inversion was found in Batumi at a concentration of 4.14 per cent in the year 1935, and in 1936 this frequency fell to 0.45 per cent. The difference between these frequencies amounts to 3.69 per cent, the error equals 1.0 per cent.

Three cases of endemic confinement of chromosome mutations were found in *D. melanogaster*. The CIIIR S inversion was found in Samarkand exclusively, CIIIR Kl – in Kutaissi only, CIIR B – in Batumi only. In these three centres there are endemic chromosome mutations, the future life history of which may lead either to destruction, or wide spreading.

The extensive diffusion or definite chromosome mutations is a consequence of successive crossing of migrant individuals. However, this diffusion does not overcome the conditions of isolation of extensive regions

present. Owing to this chromosome mutations show definite geographical regularities in their distribution throughout the area of the species. The CIIIR G inversion in *D. melanogaster* was found in North Caucasus, and crossing the Caspian Sea we found it also in Central Asia, but not in Kazakhstan (Alma-Ata). The CIIIR K2 inversion was also missing in Kazakhstan, but it spreads throughout Central Asia. On the other side of the Caspian Sea we found that in Azerbaidzhan the course taken by these inversions divides. CIIIR G spreads over North Caucasus and CIIIR K2 – Transcaucasus.

Finally, of all the areas studied the CIIL-CIIR K inversion was found only in Transcaucasus. It becomes thus clear that the main range of the Caucasian mountains represents a barrier in the distribution of these mutations.

Preliminary experiments on the viability and fertility of homozygous and heterozygous aberrations in nature have not brought out a single fact which would point out an advantage over normal wild forms.

All the inversions studied were viable in homozygous condition. It has been shown that the CIIL-CIIR K inversion is completely recessive in heterozygous condition. Females heterozygous for the inversion do not lay inviable eggs. This circumstance shows that abnormal chromosomes caused by single crossing-over between an inversion and a normal chromosome, do not enter the gametes. An oriented reduction evidently takes place in this case.

Inversions which permeate the populations are, therefore, subject to negative selection. It is difficult to say whether positive selection takes place. The possibility is not excluded that these inversions are selectively indifferent changes. In this case the multiplication of a singly arisen mutation, its gradual penetration and distribution throughout extensive areas, may take place independently of selection. The overwhelming majority of singly arisen mutations perish under the effect of the automatic genetic processes which thus act as a uniformizer. However, some mutations multiply and permeate endemic centers.

Their subsequent fate is determined by the incompleteness of isolation since each species acts potentially as a unit. Nevertheless many mutations from endemic centers perish after having multiplied and spread over certain areas. Only in rarest instances do chromosome mutations gradually overcome the barring action of incomplete isolation invading thus one population after another. Intercrossing causes successive panmixis which in the long run takes in the whole species.

This process does not in reality take place independently of selection, for should we deal even with a completely indifferent inversion, selection affects the fate of the latter by acting on the gene material it carries.

The study concerning the relation existing between the structural changes of the chromosomes, the automatic genetic processes and selection has revealed the fact that a limited number of mutations have permeated whole species or extensive areas (the CIIR-CIIL inversion is found in North America and Georgia), while the frequency of these changes varies strongly with the population.

6. The investigation of *D. obscura* 2 and *D. obscura* 3 present pictures of intrachromosomal variability which differ from those of *D. melanogaster* or *D. funebris*.

Firstly, not only inversions but also other rearrangements have been found here (intrachromosomal displacements). Secondly, populations of these species are exceptionally richly permeated with mutations. Individuals were found which carried simultaneously eight chromosomal mutations (4 inversions, 2 rearrangements and 2 complex aberrations)

which were connected with 20 breaks and corresponding exchanges of chromatin. This great number of rearrangements was found in populations which simultaneously carried many inversions in the same chromosomes. The variability of the chromosome structure, of these species is so great, that the karyotype of many individuals, collected in nature, recall the type in intraspecific hybrids obtained from combining chromosomal elements, which were separated by a long period of evolution.

7. Our results render it possible to present the following picture regarding to intraspecific chromosome variability and its significance for the evolution of the species.

A. The relative frequencies for different kinds of changes in chromosome structure, as they are turned out by the mutation process or as we find them in populations, are quite different. Observations have shown that in nature only inversions and intrachromosomal displacements (the latter with far lower frequency) are present in populations; not a single translocation was found, though they usually occur more frequently than other type of chromosome mutations.

Though it is perfectly clear that translocations must arise in nature along with the other mutations and that they must partake in the evolution of the karyotype, they are, nevertheless, found in natural populations at an incomparably lower rate than inversions and other rearrangements. The reason for this evidently lies in the action of selection. Inversions are not subject to selection. Intrachromosomal displacements probably spread only in populations saturated with inversions, for the latter in suppressing crossing-over avert the deleterious effects that the separation of complementary constituents of a displacement would produce, in case of interchange with a normal chromosome. Translocations cause in the majority of cases, non-disjunction of the components at gametogenesis, and they must be, therefore, subject to negative selection in the heterozygotes.

B. It is clear, on the other hand, that even non-adaptive chromosome mutations, similar to inversions, may play a role in the fission of species. Every aberration involving a given set of genes, acts as a separate accumulator for mutational changes. The mutations arising, for instance, in inversions do not separate from them; if the frequency, with which an inversion is present in the population, is low, then the gene content of every chromosome carrying this inversion will remain isolated, for there will be no interchange with the normal chromosome, but if the frequency rises homozygotes for their inversion will appear, and crossing-over is obtained. This represents as peculiar type of isolation of a part of the genotype. Thus, the evolving karyotype is able to split up the biological unit that species represents.

If dominant genes arise within the aberration or, if there is the position effect, the aberration undergoes selection in heterozygous condition.

C. The role played by aberrations in the evolution of the karyotype and in the divergence of the species is different for various groups of mutations. The evolution of the karyotype is founded on a successive inclusion of separate mutations. This evolution is not necessarily connected with species adaptation and species divergence. The phylogeny of the karyotype reflects the peculiarities of chromosome variability of a population. For instance, inversions that are the most widely spread have the greater chance to be incorporated in the evolution of the karyotype. In the fission of species the chromosome aberrations play an important role where they coincide with physiological isolation. Of the

greatest importance are in this connection the aberrations, which underlie strong negative selection in heterozygous condition, whereas they do not exhibit those negative properties in homozygous condition. This refers specially to translocations. Even the presence of a single translocation in homozygous condition (not of the complex type) in a population may represent a physiological barrier preventing the mixture of this population with any other not carrying this translocation.

The investigations of intraspecific variability of chromosomes constitute an exclusive method for the study of some of the evolutionary characteristics of organisms. Intrinsic features of species evolution remain concealed to direct and methodical observation. Though it is impossible to draw a complete picture for the gene mutations of a population, it is yet possible to do it in respect to chromosomal changes, for we are in a position to follow them up with great accuracy. Observations carried on over a period of years, decades and even centuries, and based on an exact valuable and irreproachable method in the field of cytogenetic investigation, will enable us to trace a detailed picture of the evolution of the karyotypes of organisms.

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